



## CYP17A1 gene

cytochrome P450 family 17 subfamily A member 1

### Normal Function

The *CYP17A1* gene provides instructions for making a member of the cytochrome P450 enzyme family. Like other cytochrome P450 enzymes, CYP17A1 is involved in the formation (synthesis) of steroid hormones. This group of hormones includes sex hormones such as testosterone and estrogen, which are needed for normal sexual development and reproduction; mineralocorticoids, which help regulate the body's salt and water balance; and glucocorticoids, which are involved in maintaining blood sugar levels and regulating the body's response to stress.

Steroid hormones are synthesized through a series of chemical reactions. The CYP17A1 enzyme performs two important reactions in this process. The enzyme has 17 alpha( $\alpha$ )-hydroxylase activity, converting pregnenolone to 17-hydroxypregnenolone and progesterone to 17-hydroxyprogesterone. These hormone precursors are further processed to produce glucocorticoids and sex hormones. CYP17A1 also has 17,20-lyase activity, which converts 17-hydroxypregnenolone to dehydroepiandrosterone (DHEA). This reaction is integral to the production of sex hormones.

### Health Conditions Related to Genetic Changes

#### 17 alpha-hydroxylase/17,20-lyase deficiency

Dozens of mutations in the *CYP17A1* gene have been found to cause 17 $\alpha$ -hydroxylase/17,20-lyase deficiency. This condition affects the function of certain hormone-producing glands, leading to high blood pressure (hypertension) and abnormal sexual development. Mutations associated with this condition reduce or eliminate both 17 $\alpha$ -hydroxylase and 17,20-lyase activity. Reduction of these activities leads to partial 17 $\alpha$ -hydroxylase/17,20-lyase deficiency, while total loss of these activities leads to the more severe form of the disorder known as complete 17 $\alpha$ -hydroxylase/17,20-lyase deficiency.

Without 17 $\alpha$ -hydroxylase activity, pregnenolone and progesterone are not converted to 17-hydroxypregnenolone or 17-hydroxyprogesterone, impairing production of glucocorticoids. Instead pregnenolone and progesterone are processed to form mineralocorticoids. An excess of these salt-regulating hormones leads to hypertension and low levels of potassium in the blood (hypokalemia).

A loss of 17,20-lyase activity impairs sex hormone production. In females, a lack of female sex hormones disrupts development of internal reproductive organs (the ovaries and uterus) and secondary sex characteristics, such as breasts and menstrual periods. In chromosomal males (individuals with an X and a Y chromosome), a lack of male sex hormones leads to abnormal development of external genitalia. Depending on the severity of the condition, these affected individuals can have abnormal male genitalia, genitalia that do not look clearly male or clearly female (ambiguous genitalia), or characteristically female genitalia.

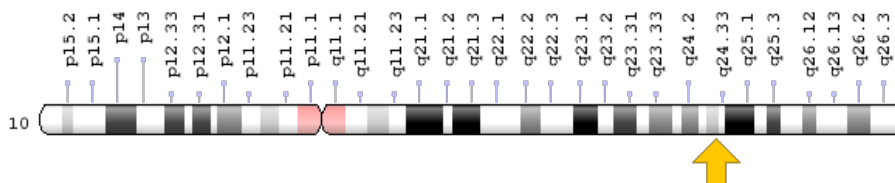
#### other disorders

A small number of *CYP17A1* gene mutations have been found to cause isolated 17,20-lyase deficiency, which is characterized by abnormal sexual development without hypertension or hypokalemia. These mutations alter a region of the *CYP17A1* protein that plays a role in the enzyme's 17,20-lyase function but not its 17 $\alpha$ -hydroxylase function. As a result, 17,20-lyase activity is severely reduced but 17 $\alpha$ -hydroxylase activity is normal. As in 17 $\alpha$ -hydroxylase/17,20-lyase deficiency (described above), impairment of 17,20-lyase activity disrupts sex hormone production, leading to abnormal development of internal or external reproductive organs and delayed or absent puberty in affected individuals.

### Chromosomal Location

Cytogenetic Location: 10q24.32, which is the long (q) arm of chromosome 10 at position 24.32

Molecular Location: base pairs 102,830,531 to 102,837,533 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- 17-alpha-hydroxyprogesterone aldolase
- CPT7
- CYP17

- CYPXVII
- cytochrome P450 17A1
- cytochrome P450-C17
- cytochrome p450 XVIIA1
- cytochrome P450, family 17, subfamily A, polypeptide 1
- cytochrome P450, subfamily XVII (steroid 17-alpha-hydroxylase), adrenal hyperplasia
- cytochrome P450c17
- P450C17
- S17AH
- steroid 17-alpha-hydroxylase/17,20 lyase precursor
- steroid 17-alpha-monooxygenase

## **Additional Information & Resources**

### Educational Resources

- Biochemistry (fifth edition, 2002): Steroid Hormones  
[https://www.ncbi.nlm.nih.gov/books/NBK22339/#\\_A3657\\_](https://www.ncbi.nlm.nih.gov/books/NBK22339/#_A3657_)
- Endocrinology: An Integrated Approach (2001): Sexual Differentiation of the External Genitalia  
[https://www.ncbi.nlm.nih.gov/books/NBK29/#\\_A1024\\_](https://www.ncbi.nlm.nih.gov/books/NBK29/#_A1024_)
- Endotext (2013): Congenital Adrenal Hyperplasia  
<https://www.ncbi.nlm.nih.gov/books/NBK278953/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CYP17A1%5BTIAB%5D%29+OR+%28cytochrome+P450+family+17+subfamily+A+member+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

### OMIM

- CYTOCHROME P450, FAMILY 17, SUBFAMILY A, POLYPEPTIDE 1  
<http://omim.org/entry/609300>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_CYP17A1.html](http://atlasgeneticsoncology.org/Genes/GC_CYP17A1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=CYP17A1%5Bgene%5D>
- HGNC Gene Family: Cytochrome P450 family 17  
<http://www.genenames.org/cgi-bin/genefamilies/set/1008>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=2593](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2593)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1586>
- UniProt  
<http://www.uniprot.org/uniprot/P05093>

## **Sources for This Summary**

- OMIM: CYTOCHROME P450, FAMILY 17, SUBFAMILY A, POLYPEPTIDE 1  
<http://omim.org/entry/609300>
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- Van Den Akker EL, Koper JW, Boehmer AL, Themmen AP, Verhoef-Post M, Timmerman MA, Otten BJ, Drop SL, De Jong FH. Differential inhibition of 17alpha-hydroxylase and 17,20-lyase activities by three novel missense CYP17 mutations identified in patients with P450c17 deficiency. *J Clin Endocrinol Metab.* 2002 Dec;87(12):5714-21.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12466376>

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